

**DUKE UNIVERSITY MEDICAL CENTER**

**CURRICULUM VITAE**

**for  
Permanent Record  
and the  
Appointments and Promotions Committee**

**Name:** Huntington Faxon Willard, Ph.D.

**Primary academic appointment:**

**Primary academic department (not DUAP):** Department of Molecular Genetics and Microbiology

**Secondary appointment (if any) – (department):**

**Present academic rank and title (if any):** Professor with Tenure (pending)

**Date and rank of first Duke Faculty appointment:** January 1, 2003  
(Instructor Temporary)

**Medical Licensure:** Not Applicable

**Specialty certifications and dates:** Not Applicable

<b>Education:</b>	<b>Institution</b>	<b>Date</b>	<b>Degree</b>
High School	Belmont Hill School	1971	Diploma
College	Harvard University	1975	A.B.
Graduate or Professional School	Yale University Department of Human Genetics	1979	Ph.D.

**Scholarly societies:**

American Society of Human Genetics  
American Society of Gene Therapy  
Human Genome Organization (HUGO)

**Professional training and academic career:**

<b>Institution</b>	<b>Position/Title</b>	<b>Dates</b>
John Hopkins University Division of Medical Genetics School of Medicine	Fellow	1979-1981
University of Toronto Departments of Medical Genetics and Medical Biophysics	Assistant Professor	1982-1987
University of Toronto Departments of Medical Genetics and Medical Biophysics	Associate Professor	1987-1989
Stanford University Department of Genetics	Associate Professor	1989-1992
Case Western Reserve University Department of Genetics	Henry Willson Payne Professor and Chairman	1992-2001
Case Western Reserve University Department of Genetics	Professor	2001-2003
Case Western Reserve University/ University Hospitals of Cleveland	Director, Center for Human Genetics	1992-2002
University Hospitals of Cleveland The Research Institute	Director and President	1999-2002
Duke University Institute for Genome Sciences and Policy	Director and Vice Chancellor	2003-

**Administrative and professional experience:**Academic and Professional Positions

2003 -	Director, Institute for Genome Sciences and Policy, Duke University
1999 - 2003	Director and President, The Research Institute of University Hospitals of Cleveland
1992 - 2002	Director, Center for Human Genetics, University Hospitals of Cleveland
1992 - 2001	Chairman, Department of Genetics, Case Western Reserve University
1993 - 2001	Member, Council of Basic Science Chairs, Case Western Reserve University School of Medicine (President, 1994-1997)
1994 - 2001	Member, Council of Clinical Chairs, Case Western Reserve University School of Medicine
1993 - 2002	Founder and Chairman, University Genetics, Inc. (Professional Practice Plan for Genetics faculty at University Hospitals of Cleveland)

#### American Society of Human Genetics

2000 - 2003	Board of Directors, American Society of Human Genetics (President, 2001)
1997 – 2001	Awards Committee, American Society of Human Genetics
1994 - 1996	Board of Directors, American Society of Human Genetics
1990 - 1994	Program Committee, American Society of Human Genetics (Chairman, 1994)

#### National Institutes of Health

1999 – 2002	Mammalian Genetics Study Section, National Institutes of Health (Chairman, 2000-2002)
1987 - 1991	NICHD Mental Retardation Research Committee, National Institutes of Health (Chairman, 1990-1991)

#### Other Scientific Review Committees

1997 – present	Scientific Review Board (Genetics), Howard Hughes Medical Institute
1992 - 1998	Basil O'Connor Advisory Committee, March of Dimes Birth Defects Foundation

#### Other National and International Organizations

2002 – 2005	Member, Excellence in Science Award Committee, Federation of American Societies for Experimental Biology (FASEB)
2000 – 2001	Member, Committee on Understanding the Biology of Sex and Gender Differences, Institute of Medicine
1997 – 2000	Member, Council of Academic Societies, American Association of Medical Colleges (AAMC)
1996 - 1999	Councilor, Association of Professors of Human/Medical Genetics
1992 - 1995	Co-editor, Genetic Constitution of the Human X Chromosome, Human Gene Mapping Workshops
1993 - 1995	Human Genome Mapping Committee, Human Genome Organization (HUGO)
1988 - 1991	Committee on Genetic Constitution of the Human X Chromosome, International Workshop on Human Gene Mapping
1982 - 1989	Committee on Human Gene Mapping Using Recombinant DNA Techniques, International Workshop on Human Gene Mapping

#### Scientific Journals

1991 - present	Co-Founder and Executive Editor, <i>Human Molecular Genetics</i>
1990 – 1999	Editor, <i>Chromosoma</i>
1987 - 1988	Editorial Board, <i>Genomics</i>
1988 - 1991	Subject Area Editor, <i>Genomics</i>
1989 - 1991	Associate Editor, <i>The American Journal of Human Genetics</i>
1984 - 1989	Editorial Board, <i>Cytogenetics and Cell Genetics</i>

#### **Publications:**

##### **Refereed journals**

1. Latt SA, Stetten G, Juergens LA, **Willard HF**, Scher CD (1975). Recent developments in the detection of DNA synthesis by 33258 Hoechst fluorescence. *J. Histochem. Cytochem.* 23:493-505.

2. **Willard HF**, Latt SA (1976). Analysis of DNA replication in human X chromosomes by fluorescence microscopy. *Am. J. Hum. Genet.* 28:213-227.
3. Latt SA, **Willard HF**, Gerald PS (1976). BrdU-33258 Hoechst analysis of DNA replication in human lymphocytes with supernumerary or structurally abnormal X chromosomes. *Chromosoma* 57:135-153.
4. **Willard HF**, Ambani LM, Hart AC, Mahoney MJ, Rosenberg LE (1976). Rapid prenatal and postnatal detection of inborn errors of propionate, methylmalonate and cobalamin metabolism: a sensitive assay using cultured cells. *Hum. Genet.* 34:277-283.
5. **Willard HF** (1977). Tissue-specific heterogeneity in DNA replication patterns of human X chromosomes. *Chromosoma* 61:61-73.
6. Mellman IS, Youngdahl-Turner P, **Willard HF**, Rosenberg LE (1977). Intracellular binding of radioactive hydroxocobalamin to cobalamin-dependant apoenzymes in rat liver. *Proc. Natl. Acad. Sci. USA* 74:916-920.
7. **Willard HF**, Rosenberg LE (1977). Inherited deficiencies of human methylmalonyl CoA mutase activity: reduced affinity of mutant apoenzyme for adenosylcobalamin. *Biochem. Biophys. Res. Comm.* 78:927-934.
8. **Willard HF**, Mellman IS, Rosenberg LE (1978). Genetic complementation among inherited deficiencies of methylmalonyl CoA activity: evidence for a new class of human cobalamin mutant. *Am. J. Hum. Genet.* 30:1-13.
9. Mellman IS, **Willard HF**, Rosenberg LE (1978). Cobalamin binding and cobalamin-dependent enzyme activity in normal and mutant human fibroblasts. *J. Clin. Invest.* 62:952-960.
10. **Willard HF**, Rosenberg LE (1979). Inborn errors of cobalamin metabolism: effect of cobalamin supplementation in culture on methylmalonyl CoA mutase activity in normal and mutant human fibroblasts. *Biochem. Genet.* 17:57-75.
11. Mellman IS, **Willard HF**, Youngdahl-Turner P, Rosenberg LE (1979). Cobalamin coenzyme synthesis in normal and mutant human fibroblasts: evidence for a processing enzyme activity deficient in *cbl C* cells. *J. Biol. Chem.* 254:11847-11853.
12. Wolf B, **Willard HF**, Rosenberg LE (1980). Kinetic analysis of genetic complementation in heterokaryons of propionyl CoA carboxylase-deficient human fibroblasts. *Am. J. Hum. Genet.* 32:16-25.
13. **Willard HF**, Rosenberg LE (1980). Interactions of methylmalonyl CoA mutase from human fibroblasts with adenosylcobalamin and methylmalonyl CoA: evidence for non-equivalent active sites. *Arch. Biochem. Biophys.* 200:130-139.
14. **Willard HF**, Breg WR (1980). Human X chromosomes: synchrony of DNA replication in diploid and triploid fibroblasts with multiple active or inactive X chromosomes. *Somat. Cell Genet.* 6:187-198.
15. **Willard HF**, Rosenberg LE (1980). Inherited methylmalonyl CoA mutase apoenzyme deficiency in human fibroblasts: evidence of allelic heterogeneity, genetic compounds, and codominant expression. *J. Clin. Invest.* 65:690-698.
16. Schmeckpeper BJ, **Willard HF**, Smith KD (1981). Isolation and characterization of cloned human DNA fragments carrying reiterated sequences common to both autosomes and the X chromosome. *Nucl. Acids Res.* 9:1853-1872.
17. Fenton WA, Hack A, **Willard HF**, Gertler A, Rosenberg LE (1982). Purification and properties of methylmalonyl CoA mutase from normal human liver. *Arch. Biochem. Biophys.* 214:815-823.

18. **Willard HF**, Smith KD, Sutherland J (1983). Isolation and characterization of a major tandem repeat family from the human X chromosome. *Nucl. Acids Res.* 11:2017-2033.
19. **Willard HF**, Holmes MT (1984). Sensitive and dependable assay for distinguishing hamster and human X-linked steroid sulfatase activity in somatic cell hybrids. *Hum. Genet.* 66:272-275.
20. Worton RG, Duff C, Sylvester JE, Schmickel RD, **Willard HF** (1984). Duchenne muscular dystrophy involving translocation of the dmd gene next to ribosomal RNA genes. *Science* 224:1447-1449.
21. Caccia N, Kronenberg M, Saxe D, Haars R, Bruns G, Goverman J, Malissen M, **Willard HF**, Yoshikai J, Simon M, Hood L, Mak T (1984). The T cell receptor  $\beta$  chain genes are located on chromosome 6 in mouse and chromosome 7 in humans. *Cell* 37:1091-1099.
22. Korneluk RG, Quan F, Lewis W, Guise KS, **Willard HF**, Holmes MT, Gravel RA (1984). Isolation of human fibroblast catalase cDNA clones: Sequence of clones derived from spliced and unspliced mRNA. *J. Biol. Chem.* 259:13819-13823.
23. Rubin JS, Prideaux VR, **Willard HF**, Dulhanty AM, Whitmore GF, Bernstein A (1985). Molecular cloning and chromosomal localization of DNA sequences associated with a human DNA repair gene. *Mol. Cell Biol.* 5:398-405.
24. O'Dowd B, Quan F, **Willard HF**, Korneluk RG, Lowden JA, Gravel RA, Mahuran DJ (1985). Isolation and characterization of cDNA clones encoding the  $\beta$ -subunit of human  $\beta$ -hexosaminidase. *Proc. Natl. Acad. Sci. USA* 82:1184-1188.
25. Ingle C, Williamson R, de la Chapelle A, Herva RR, Haapala K, Bates G, **Willard HF**, Davies KE (1985). Mapping DNA sequences in a human X chromosome deletion which extends across the region of the Duchenne muscular dystrophy mutation. *Am. J. Hum. Genet.* 37:451-472.
26. **Willard HF** (1985). Chromosome-specific organization of human alpha satellite DNA. *Am. J. Hum. Genet.* 37:524-532.
27. Wolfe J, Darling SM, Erickson RP, Craig I, Buckle V, Rigby P, **Willard HF**, Goodfellow P (1985). Isolation and characterization of an alphoid centromeric repeat family from the human Y chromosome. *J. Mol. Biol.* 182:477-485.
28. Bakker E, Hofker MH, Goor N, Mandel JL, Davis KE, Kunkel LM, **Willard HF**, Fenton WA, Sandkuyl L, Majoor-Krakauer D, v. Essen AJ, Jahoda MGJ, Sachs ES, Van Ommen GJB, Pearson PL (1985). Prenatal diagnosis and carrier-detection of Duchenne muscular dystrophy with closely linked RFLPs. *Lancet* 2:655-658.
29. Waye JS, **Willard HF** (1985). Chromosome-specific alpha satellite DNA: Nucleotide sequence analysis of the 2.0 kilobasepair repeat from the human X chromosome. *Nucl. Acids Res.* 12:2731-2743.
30. Michalopoulos EE, Bevilacqua PJ, Stokoe N, Powers VE, **Willard HF**, Lewis WH (1985). Molecular analysis of gene deletion in aniridia-Wilms tumor association. *Hum. Genet.* 70:157-162.
31. **Willard HF**, Meakin S, Tsui LC, Breitman M (1985). Assignment of the human gamma crystallin multigene family to chromosome 2. *Somat. Cell Mol. Genet.* 11:511-516.
32. **Willard HF**, Goss SJ, Holmes MT, Munroe DL (1985). Regional localization of the phosphoglycerate kinase gene and pseudogene on the human X chromosome and assignment of a related DNA sequence to chromosome 19. *Hum. Genet.* 71:138-143.
33. Schmeckpeper B, Davis J, **Willard HF**, Smith K (1985). An anonymous single-copy X chromosome RFLP for DXS72 from Xq13-Xq22. *Nucl. Acids Res.* 13:5724.

34. **Willard HF**, Riordan JR (1985). Assignment of the gene for myelin proteolipid protein to the X chromosome: implications for X-linked inherited disorders of myelin. *Science* 230:940-942.
35. Lewis WH, Goguen JM, Powers VE, **Willard HF**, Michalopoulos EE (1985). Gene order on the short arm of human chromosome 11: regional assignment of the LDHA gene distal to catalase in two translocations. *Hum. Genet.* 71:249-253.
36. Lamhonwah AM, Barankiewicz TJ, **Willard HF**, Mahuran DJD, Quan F, Gravel RA (1986). Isolation of cDNA clones coding for the alpha and beta chains of human propionyl CoA carboxylases: chromosomal assignments and DNA polymorphisms associated with PCCA and PCCB genes. *Proc. Natl. Acad. Sci. USA* 83:4864-4868.
37. Buchwald M, Zsiga M, Markiewicz D, Plavsic N, Kennedy D, Zengerling S, **Willard HF**, Tsipouras P, Schmiegelow K, Schwartz M, Eiberg H, Mohr J, Donis-Keller H, Tsui L-C (1986). Linkage of cystic fibrosis to the pro  $\alpha 2$  (I) collagen gene, COL1A2, on chromosome 7. *Cytogenet. Cell. Genet.* 41:234-239.
38. Korneluk RG, Mahuran DJ, Neote K, Klavins MH, O'Dowd BF, Tropak M, **Willard HF**, Anderson MJ, Lowden JA, Gravel RA (1986). Isolation of cDNA clones coding for the  $\alpha$  subunit of human  $\beta$ -hexosaminidase: extensive homology between the  $\alpha$  and  $\beta$  subunits and studies on Tay-Sachs disease. *J. Biol. Chem.* 261:8407-8413.
39. Williams BRG, Saunders M, **Willard HF** (1986). The interferon-induced human 2-5A synthetase gene is on chromosome 12. *Somat. Cell Mol. Genet.* 12:403-408.
40. **Willard HF**, Waye JS, Skolnick MH, Schwartz CE, Powers VE, England SB (1986). Restriction fragment length polymorphisms at the centromeres of human chromosomes using chromosome-specific alpha satellite DNA: implications for development of centromere-based genetic linkage maps. *Proc. Natl. Acad. Sci. USA* 83:5611-5615.
41. Squire J, Dryja TP, Dunn JJ, Goddard A, Hoffman T, Musarella M, **Willard HF**, Becker AJ, Gallie BL, Phillips RA (1986). Cloning of the esterase D gene: a polymorphic gene probe closely linked to the retinoblastoma locus on chromosome 13. *Proc. Natl. Acad. Sci. USA* 83:6573-6577.
42. Glaser T, Lewis WH, Bruns GAP, Watkins PC, Roger LE, Shows TB, Powers VE, **Willard HF**, Goguen JM, Simola KOJ, Housman DE (1986). Beta subunit of follicle stimulating hormone is deleted in patients with aniridia and Wilm's tumor: a further definition of the WAGR locus. *Nature* 321:882-887.
43. Waye JS, **Willard HF** (1986). Structure, organization, and sequence of alpha satellite DNA from human chromosome 17: evidence for evolution by unequal crossing-over and an ancestral pentamer repeat shared with the human X chromosome. *Mol. Cell. Biol.* 6:3156-3165.
44. Waye JS, **Willard HF** (1986). Molecular analysis of a deletion polymorphism in alpha satellite of human chromosome 17: evidence of homologous unequal crossing-over and subsequent fixation. *Nucl. Acids Res.* 14:6915-6927.
45. Cooke NE, **Willard HF**, David EV, George DL (1986). Direct regional assignment of the gene for vitamin D-binding protein (Gc-globulin) to human chromosome 4q11-q13 and identification of associated DNA polymorphisms. *Hum. Genet.* 73:225-229.
46. O'Dowd BF, Klavins MH, **Willard HF**, Gravel RA, Lowden A, Mahuran DJ (1986). Molecular heterogeneity in the infantile and juvenile forms of Sandhoff disease. *J. Biol. Chem.* 261:12680-12685.

47. Durfy SJ, Clark SC, Williams BRG, **Willard HF** (1986). RFLP detected by an X-linked cDNA encoding erythroid-potentiating activity / tissue inhibitor of metalloproteinase (EPA/TIMP). *Nucl. Acids Res.* 14:9226.
48. Waye JS, England SB, **Willard HF** (1987). Genomic organization of alpha satellite DNA on human chromosome 7: evidence for two distinct alphoid domains on a single chromosome. *Mol. Cell. Biol.* 7:349-356.
49. Bell DR, Trent JM, **Willard HF**, Riordan JR, Ling V (1987). Chromosomal location of human P-glycoprotein gene sequences. *Cancer Genet. Cytogenet.* 25:141-148.
50. Wu J-S, Riordan JR, **Willard HF**, Milner R, Kidd KK (1987). MSP RFLP for X-linked proteolipid protein gene (PLP) identified with either rat or human PLP cDNA clone. *Nucl. Acids Res.* 15:1882.
51. Barker D, Wright E, Nguyen K, Cannon L, Fain P, Goldgar D, Bishop DT, Carey J, Baty B, Kivlin J, **Willard HF**, Waye JS, Greig G, Leinwand L, Nakamura Y, O'Connell P, Leppert M, Lalouel JM, White R, Skolnick M (1987). Gene for von Recklinghausen neurofibromatosis is in the pericentromeric region of chromosome 17. *Science* 236:1100-1102.
52. Waye JS, Creeper LA, **Willard HF** (1987). Organization and evolution of alpha satellite DNA from human chromosome 11. *Chromosoma* 95:182-188.
53. **Willard HF**, Waye JS (1987). Hierarchical order in chromosome-specific human alpha satellite DNA. *Trends in Genetics* 3:192-198.
54. Mengle-Gaw L, **Willard HF**, Smith CIE, Hammarstrom L, Fischer P, Sherrington P, Lucas G, Thompson PW, Baer R, Rabbitts TH (1987). Human T-Cell tumours containing chromosome 14 inversion or translocation with breakpoints proximal to immunoglobulin joining regions at 14q32. *EMBO J.* 6:2273-2280.
55. **Willard HF**, Waye JS (1987). Chromosome-specific subsets of human alpha satellite DNA: Analysis of sequence divergence within and between chromosomal subsets and evidence for an ancestral pentameric repeat. *J. Mol. Evol.* 25:207-214.
56. Durfy SJ, **Willard HF** (1987). Molecular analysis of a polymorphic domain of X chromosome alpha satellite DNA. *Am. J. Hum. Genet.* 41:391-401.
57. MacLennan DH, Brandl CJ, Champaneria S, Holland PC, Powers VE, **Willard HF** (1987). Fast-twitch and slow-twitch / cardiac  $Ca^{2+}$  ATPase genes map to human chromosomes 16 and 12. *Somat. Cell Mol. Genet.* 13:341-346.
58. O'Dowd B, Neote K, Munroe DLG, Gravel RA, Mahuran D, **Willard HF** (1987). PstI RFLP in the human hexosaminidase (*HEXB*) gene on chromosome 5. *Nucl. Acids Res.* 15:3194.
59. Waye JS, Durfy SJ, Pinkel D, Kenwrick S, Patterson M, Davies KE, **Willard HF** (1987). Chromosome-specific alpha satellite DNA from human chromosome 1: Hierarchical structure and genomic organization of a polymorphic domain spanning several hundred kilobasepairs of centromeric DNA. *Genomics* 1:43-51.
60. Waye JS, **Willard HF** (1987). Nucleotide sequence heterogeneity of alpha satellite repetitive DNA: a survey of alphoid sequences from different human chromosomes. *Nucl. Acids Res.* 15:7549-7580.
61. Waye JS, Greig GM, **Willard HF** (1987). Detection of novel centromeric polymorphisms associated with alpha satellite DNA from human chromosome 11. *Hum. Genet.* 77:151-156.

62. Barker D, Green P, Knowlton R, Schumm J, Lander E, Oliphant A, **Willard HF**, Akots G, Brown V, Gravius T, Helms C, Nelson C, Parker C, Rediker K, Watt D, Weiffenbach B, Donis-Keller H (1987). A genetic linkage map of 63 chromosome 7 DNA markers. *Proc. Natl. Acad. Sci. USA* 84:8006-8010.
63. Barker D, Wright E, Nguyen K, Cannon L, Fain P, Goldgar D, Bishop DT, Carey J, Kivlin J, **Willard HF**, Nakamura Y, O'Connell P, Leppert P, White R, Skolnick M (1987). A genomic search for linkage of NF to RFLP's. *J. Med. Genet.* 24:536-538.
64. **Willard HF**, Greig GM, Powers VE, Waye JS (1987). Molecular organization and haplotype analysis of centromeric DNA from human chromosome 17: implications for linkage in neurofibromatosis. *Genomics* 1: 368-373.
65. Vogelstein B, Fearon ER, Hamilton SR, Preisinger AC, **Willard HF**, Michelson AM, Riggs AD, Orkin S (1987). Clonal analysis using recombinant DNA probes from the human X chromosome. *Cancer Res.* 47:4806-4813.
66. Brown CJ, **Willard HF** (1987). MspI RFLP detected with chromosome-walk clone pXUT23-SE3.2L from DXS16 in Xp22.1-22.3. *Nucl. Acids Res.* 15:9614.
67. Fain PR, Barker DF, Goldgar DE, Wright E, Nguyen K, Carey J, Johnson J, Kivlin J, **Willard HF**, Mathew C, Ponder B, Skolnick M (1987). Genetic analysis of NF1: identification of close flanking markers on chromosome 17. *Genomics* 1:340-345.
68. Spence JF, Perciaccante RG, Greig GM, **Willard HF**, Ledbetter DH, Hejtmancik JF, Pollack MS, O'Brien WE, Beaudet AL (1988). Uniparental disomy as a mechanism for human genetic disease. *Am. J. Hum. Genet.* 42:217-226.
69. Worton RG, Sutherland J, Sylvester JE, **Willard HF**, Bodrug S, Dube I, Duff C, Kean V, Ray PN, Schmickel RD (1988). Human ribosomal RNA genes: orientation of the tandem array and conservation of the 5' end. *Science* 239:64-68.
70. Tsiouras P, Schwartz RC, Phillips JA III, **Willard HF** (1988). A centromere based linkage group on the long arm of chromosome 17. *Cytogenet. Cell Genet.* 47:109-111.
71. Waye JS, Mitchell AR, **Willard HF** (1988). Organization and genomic distribution of '82H' alpha satellite DNA: Evidence for a low-copy or single-copy alphoid domain located on human chromosome 14. *Hum. Genet.* 78:27-32.
72. Waye JS, Gravel RA, **Willard HF** (1988). Two PstI RFLPs in the PCCB gene on the long arm of chromosome 3. *Nucl. Acids Res.* 16:2362.
73. Allore R, O'Hanlon D, Price R, Neilson K, **Willard HF**, Cox D, Marks A, Dunn RJ (1988). Gene encoding the  $\beta$  subunit of S100 protein is on chromosome 21: implications for Down Syndrome. *Science* 239:1311-1313.
74. Lubahn DB, Joseph DR, Sullivan DM, **Willard HF**, French FS, Wilson EM (1988). Cloning of human androgen receptor cDNA and localization to the X chromosome. *Science* 240:327-330.
75. Otulakowski G, Robinson BH, **Willard HF** (1988). The gene for lipoamide dehydrogenase maps to human chromosome 7. *Somat. Cell Mol. Genet.* 14:411-414.
76. Mahtani MM, **Willard HF** (1988). A primary genetic map of the pericentromeric region of the human X chromosome. *Genomics* 2:294-301.
77. Devilee P, Kievits T, Waye JS, Pearson PL, **Willard HF** (1988). Chromosome-specific alpha satellite DNA: isolation and mapping of a polymorphic alphoid repeat from human chromosome 10. *Genomics* 3:1-7.



78. Mitchell GA, Looney JE, Brody LC, Steel G, Suchanek M, Engelhardt JF, **Willard HF**, Valle D (1988). Human ornithine- $\delta$ -aminotransferase: cDNA cloning and analysis of the structural gene. *J. Biol. Chem.* 263:14288-14295.
79. Brown CJ, Mahtani MM, **Willard HF** (1988). Genetic mapping of four DNA markers (DXS16, DXS43, DXS85 and DXS143) from the p22 region of the human X chromosome. *Hum. Genet.* 80:296-298.
80. Devilee P, Thierry RF, Kievits T, Kolluri R, Hopman AHN, **Willard HF**, Pearson PL, Cornelisse CJ (1988). Detection of chromosome aneuploidy in interphase nuclei from human primary breast tumours using chromosome-specific repetitive DNA probes. *Cancer Res.* 48:5825-5830.
81. Brown CJ, Sekiguchi T, Nishimoto T, **Willard HF** (1989). Regional localization of the CCG1 gene which complements the hamster cell cycle mutation BN462 to Xq11-Xq13. *Somat. Cell Mol. Genet.* 15:93-96.
82. Stephens K, Green P, Riccardi VM, Ng S, Rising M, Barker D, Darby J, Falls KM, Collins FS, **Willard HF**, Donis-Keller H (1989). Genetic analysis of eight loci tightly linked to neurofibromatosis. *Am. J. Hum. Genet.* 44:13-19.
83. Fain PR, Wright E, **Willard HF**, Stephens K, Barker DF (1989). The order of loci in the pericentric region of chromosome 17, based on evidence from physical and genetic breakpoints. *Am. J. Hum. Genet.* 44:68-72.
84. **Willard HF**, Durfy SJ, Mahtani MM, Dorkins H, Davies KE, Williams BRG (1989). Regional localization of the TIMP gene on the human X chromosome: extension of a conserved synteny and linkage group on proximal Xp. *Hum. Genet.* 81:234-238.
85. Logan C, **Willard HF**, Rommens JM, Joyner AL (1989). Chromosomal localization of the human homeo box-containing genes, *EN1* and *EN2*. *Genomics* 4:206-209.
86. Brown CJ, Goss SJ, Lubahn DB, Joseph DR, Wilson EM, French FS, **Willard HF** (1989). Androgen receptor locus on the human X chromosome: Regional localization to Xq11-12 and description of a DNA polymorphism. *Am. J. Hum. Genet.* 44:264-269.
87. Waye JS, **Willard HF** (1989). Chromosome-specificity of satellite DNAs: short and long-range organization of a diverged dimeric subset of human alpha satellite from chromosome 3. *Chromosoma* 95:275-280.
88. Brown CJ, Powers VE, Munroe DL, Sheinin R, **Willard HF** (1989). A gene on the short arm of the human X chromosome complements the murine tsA1S9 DNA synthesis mutation. *Somat. Cell Mol. Genet.* 15:173-178.
89. Waye JS, **Willard HF** (1989). Human beta satellite DNA: genomic organization and sequence definition of a class of highly repetitive tandem DNA. *Proc. Natl. Acad. Sci. USA* 86:6250-6254.
90. Waye JS, **Willard HF** (1989). Concerted evolution of alpha satellite DNA: evidence for species specificity and a general lack of sequence conservation among alphoid sequences of higher primates. *Chromosoma* 98:273-279.
91. Alitalo T, **Willard HF**, de la Chapelle A (1989). Determination of the breakpoint of 1;7 translocations in myelodysplastic syndrome by in situ hybridization using chromosome-specific alpha satellite DNA from human chromosomes 1 and 7. *Cytogenet. Cell Genet.* 50:49-53.
92. Greer WL, Mahtani MM, Kwong PC, Rubin LA, Peacocke M, **Willard HF**, Siminovitch KA (1989). Linkage studies of the Wiskott-Aldrich syndrome: polymorphisms at TIMP and the X chromosome centromere are informative markers for genetic prediction. *Hum. Genet.* 83:227-230.

93. Brown CJ, **Willard HF** (1989). Non-inactivation of a selectable human X-linked gene that complements a murine temperature-sensitive cell cycle defect. *Am. J. Hum. Genet.* 45:592-598.
94. Durfy SJ, **Willard HF** (1989). Patterns of intra- and interarray sequence variation in alpha satellite from the human X chromosome: Evidence for short range homogenization of tandemly repeated DNA sequences. *Genomics* 5:810-821.
95. Wevrick R, **Willard HF** (1989). Long-range organization of tandem arrays of alpha satellite at the centromeres of human chromosomes: High frequency array length polymorphism and meiotic stability. *Proc. Natl. Acad. Sci. USA* 86:9394-9398.
96. Lafreniere RG, Mahtani MM, **Willard HF** (1989). An X-linked Dra I RFLP recognized by cpX23 (DXS132). *Nucl. Acids Res.* 17:6754.
97. Greig GM, England SB, Bedford HM, **Willard HF** (1989). Chromosome-specific alpha satellite DNA from the centromere of human chromosome 16. *Am. J. Hum. Genet.* 45:862-872.
98. Tran-Paterson R, **Willard HF**, Letarte M (1989). The common acute lymphoblastic leukemia antigen (neutral endopeptidase-3.4.24.11) gene is located on human chromosome 3. *Cancer Genet. Cytogenet.* 42:129-134.
99. Brown CJ, **Willard HF** (1990). Localization of a gene that escapes inactivation to the X chromosome proximal short arm: implications for X inactivation. *Am. J. Hum. Genet.* 45:273-279.
100. Koch J, Kolvraa S, Hobolt N, Petersen GB, **Willard HF**, Waye JS, Gregersen N, Bolund L (1990). A case of 46,XX,r(X) (p1q1) diagnosed by in situ hybridization. *Clin. Genet.* 37:216-220.
101. Luty JA, Guo Z, **Willard HF**, Ledbetter DH, Ledbetter S, Litt M (1990). Five polymorphic microsatellite VNTRs on the human X chromosome. *Am. J. Hum. Genet.* 46:776-783.
102. Fujii J, **Willard HF**, MacLennan DH (1990). Characterization and localization to human chromosome 1 of the human fast-twitch skeletal muscle calsequestrin gene. *Somat. Cell Mol. Genet.* 16:185-189.
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#### **Non-refereed publications:**

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4. Brown CJ, **Willard HF** (1993). Molecular and genetic studies of human X chromosome inactivation. *Adv. Devel. Biol.* 2: 37-72.
5. Tyler-Smith C, **Willard HF** (1993). Mammalian chromosome structure. *Curr. Opin. Genet. Devel.* 3: 390-397.
6. **Willard HF**, Brown CJ, Carrel L, Hendrich B, Miller AP (1993). Epigenetic and chromosomal control of gene expression: molecular and genetic analysis of X chromosome inactivation. *Cold Spring Harbor Symp. Quant. Biol.* 58: 315-322.
7. **Willard HF** (1996). X chromosome inactivation and X-linked mental retardation. *Am. J. Med. Genet.* 64: 21-26.
8. **Willard HF** (1996). Chromosome manipulation: a systematic approach toward understanding human chromosome structure and function. *Proc. Natl. Acad. Sci. USA* 93: 6847-6850.
9. **Willard HF** (1996). X chromosome inactivation, *XIST*, and pursuit of the X inactivation center. *Cell* 86: 5-7.
10. Sullivan BA, Schwartz S, **Willard HF** (1996). Centromeres of human chromosomes. *Envir. Mol. Mutagen.* 28: 182-191.
11. **Willard HF**, Salz HK (1997). Dosage compensation: remodelling chromatin with RNA. *Nature* 386: 228-229.
12. Puck JM, **Willard HF** (1998). X inactivation in females with X-linked disease. *New Engl. J. Med.* 338: 291-295.
13. **Willard HF** (1998). Centromeres: the 'missing link' in the development of human artificial chromosomes. *Curr. Opin. Genet. Dev.* 8: 219-225.
14. **Willard HF** (1998). Human artificial chromosomes coming into focus. *Nature Biotechnol.* 16: 415-416.

15. Carrel L, **Willard HF** (1998). Counting on Xist [News & Views]. *Nature Genet.* 19: 211-212.
16. **Willard HF**, Hendrich BD (1999). Breaking the silence in Rett syndrome [News & Views]. *Nature Genet.* 23: 127-128.
17. **Willard HF** (2000). Artificial chromosomes coming to life [Perspectives]. *Science* 290: 1308-1309.
18. **Willard HF**, Carrel L (2001). Making sense (and antisense) of the X inactivation center [Commentary]. *Proc. Natl. Acad. Sci. USA* 98: 10025-10027.
19. **Willard HF** (2001). Neocentromeres and human artificial chromosomes: an unnatural act [Commentary]. *Proc. Natl. Acad. Sci. USA* 98: 5374-5376.
20. **Willard HF** (2002). ASHG Presidential Address – On black boxes and storytellers: lessons learned in human genetics. *Am. J. Hum. Genet.* 70: 285-296.

#### **Chapters in books:**

1. **Willard HF**, Rosenberg LE (1979). Inherited deficiencies of methylmalonyl CoA mutase activity: biochemical and genetic studies in cultured skin fibroblasts. In: *Models for the Study of Inborn Errors of Metabolism* (F.A. Hommes, ed.) Elsevier/North-Holland Biomedical Press, pp. 297-310.
2. **Willard HF** (1983). Replication of human X chromosomes in cultured fibroblasts and in somatic cell hybrids: Cytogenetic analysis and a molecular perspective. In: *'Cytogenetics of Mammalian X Chromosomes'* (A.A. Sandberg, ed.). Alan R. Liss, Inc., New York, pp. 427-448.
3. **Willard HF** (1983). Molecular and cytogenetic mapping studies of mammalian X chromosomes: Ohno's hypothesis revisited. In: *'Cytogenetics of Mammalian X Chromosomes'* (A.A. Sandberg, ed.). Alan R. Liss, Inc., New York, pp. 463-478.
4. **Willard HF** (1985). Molecular organization of repeated DNA sequences on the human Y chromosome. In: *'The Y Chromosome'*, (A.A. Sandberg, ed.), Alan R. Liss Inc., New York, pp. 125-140.
5. O'Dowd BF, Klavins MH, **Willard HF**, Gravel R, Lowden JA, Mahuran DJ (1986). Molecular heterogeneity in O-variant GM2 gangliosidosis. In: *'Enzymes of Lipid Metabolism'* (R. Massarelli and S. Gatt, eds.), Plenum Publishing Corp., NY, pp. 779-784.
6. **Willard HF**, Wevrick R, Warburton PE (1989). Human centromere structure: organization and potential role of alpha satellite DNA. In: *Aneuploidy: Mechanisms of Origin* (MA Resnick, ed.). Alan R. Liss, Inc., New York, pp. 9-18.
7. **Willard HF** (1989). The genomics of long tandem arrays of satellite DNA in the human genome. In: *Proceedings of the Sixteenth International Congress of Genetics.* *Genome* 31:737-744.
8. **Willard HF** (1990). Molecular cytogenetics of centromeres of human chromosomes. In: *Chromosomes Today*, vol. 10:47-60 (Fredga K, ed.), Unwin Hyman, London.

9. **Willard HF** (1990). Alpha and beta satellite sequences on chromosome 21: the possible role of centromere and chromosome structure in nondisjunction. In: *The Molecular Genetic Analysis of Chromosome 21* (Epstein CJ, Patterson D, eds.), Alan R. Liss, New York, pp. 39-52.
10. **Willard HF** (1994). Human genetics. In: *Introduction to Molecular Medicine*, Leder P, Clayton D, Rubinstein E (eds). New York: Scientific American Medicine, pp. 1-26.
11. **Willard HF** (1995). Sex chromosomes and X chromosome inactivation, In: *The Metabolic and Molecular Bases of Inherited Disease*, 7th ed., Scriver CR, Beaudet AL, Sly WS, Valle D (eds). New York: McGraw-Hill Publishing Co., pp. 719-737.
12. Pyeritz RE, Fearon ER, Seidman C, **Willard HF** (1995). Genetics in clinical medicine, In: MKSAP 10, Goldman L (ed). Philadelphia: American College of Physicians, pp. 1113-1150.
13. Tilghman SM, **Willard HF** (1995). Epigenetic regulation in mammals. In: *Chromatin Structure and Gene Expression* (Elgin S, ed.). Oxford: Oxford Univ. Press, pp. 197-222.
14. Warburton PE, **Willard HF** (1996). Evolution of centromeric alpha satellite DNA: molecular organization within and between human and primate chromosomes. In: *Human Genome Evolution*, Jackson M, Strachan T, Dover G, eds. Oxford: BIOS Scientific Publishers, pp. 121-145.
15. **Willard HF** (1997). X chromosome inactivation. In: *Encyclopedia of Human Biology*, 2nd edition, vol. 8. Academic Press, New York, pp. 785-789.
16. **Willard HF** (2000). The sex chromosomes and X chromosome inactivation. In: *The Metabolic and Molecular Bases of Inherited Disease*, 8<sup>th</sup> ed. Edited by Scriver CR, Beaudet AL, Sly WS, Valle D, Childs B, Vogelstein B. New York: McGraw-Hill, pp. 1191-1221.
17. **Willard HF** (2000). Chromonomics: structure and function of natural and artificial human chromosomes. In: *Current Options for the Human Genome Project*. Edited by S. Grisolia. Madrid: Fundacion BBV, pp. 253-262.

#### **Books:**

- Thompson MW, McInnes RR, **Willard HF** (1991). *Genetics in Medicine*, Fifth Edition. Philadelphia: WB Saunders Co., 500 pp.
- Nussbaum RL, McInnes RR, **Willard HF** (2001). *Genetics in Medicine*, Sixth Edition. Philadelphia: WB Saunders Co., 444 pp.

### Other published scientific reviews:

1. Skolnick MH, **Willard HF**, Menlove L (1984). Human gene mapping by recombinant DNA techniques. Seventh International Workshop on Human Gene Mapping. Cytogenet. Cell Genet. 34:210-273.
2. **Willard HF**, Skolnick MH, Pearson PL, Mandel JL (1985). Human gene mapping by recombinant DNA techniques. Eighth International Workshop on Human Gene Mapping. Cytogenet. Cell Genet. 4:360-489.
3. Kidd KK, **Willard HF**, Pearson PL, Skolnick MH, Cohen IH, Miller RL, Track RK (1987). Human DNA restriction fragment length polymorphisms (RFLPs). In: "Genetic Maps 1987" (SJ O' Brien, ed.), Cold Spring Harbor Press, New York, pp. 610-641.
4. Pearson PL, Kidd KK, **Willard HF** (1987). Human gene mapping by recombinant DNA techniques. Ninth International Workshop on Human Gene Mapping. Cytogenet. Cell Genet. 46:390-566.
5. Mandel JL, **Willard HF**, Nussbaum RL, Davies KE, Romeo G (1988). Report of the committee on the genetic constitution of the X chromosome. Update to the Ninth International Workshop on Human Gene Mapping. Cytogenet. Cell Genet. 49:107-128.
6. Kidd KK, Bowcock AM, Pearson PL, Schmidtke J, **Willard HF**, Track RK, Ricciuti R (1988). Human gene mapping by recombinant DNA techniques. Update to the Ninth International Workshop on Human Gene Mapping. Cytogenet. Cell Genet. 49:133-218.
7. Davies KE, Mandel JL, Monaco AP, Nussbaum RL, **Willard HF** (1990). Report of the committee on the constitution of the X chromosome, Human Gene Mapping 10.5: Update to the Tenth International Workshop on Human Gene Mapping. Cytogenet. Cell Genet. 55:254-313.
8. Schlessinger D, Mandel JL, Monaco AP, Nelson DL, **Willard HF** (1993). Report of the fourth international workshop on human X chromosome mapping. Cytogenet. Cell Genet. 64: 148-194.
9. **Willard HF**, Mandel JL, Monaco AP, Nelson DL, Schlessinger D (1994) Report of the committee on the genetic constitution of the X chromosome. In: *Human Gene Mapping 1993, A compendium*. (Cutichchia AJ, Pearson PL, eds.). Baltimore: Johns Hopkins University Press, pp. 656-719.
10. **Willard HF**, Cremers F, Mandel JL, Monaco AP, Nelson DL, Schlessinger D (1994). Report of the fifth international workshop on human X chromosome mapping. Cytogenet. Cell Genet. 67: 295-358.

### Editorials

**Willard HF** (1992). Centromeres -- primary constrictions are primarily complicated. Hum. Mol. Genet. 1: 667-668.

**Willard HF** (1993). Cloning of the X-linked glycerol kinase gene. Hum. Mol. Genet. 2: 95-96.

**Willard HF** (1993). The needle found! Trinucleotide repeat expansion in the Huntington's disease gene. Hum. Mol. Genet. 2: 497-498.

**Willard HF**, Davies KE (1993). Mammalian genetics: a sampler of current opinion. Curr. Opin. Genet. Devel. 3: 387-389.

**Willard HF** (2001). You say tomato and I say tomahto: human genetics and gene therapy. Mol. Therapy 4: 514.

### **Invited Editorial Review**

Cell  
Molecular Cell  
Developmental Cell  
Science  
Nature  
Nature Genetics  
Nature Neuroscience  
Proceedings of the National Academy  
of Sciences, USA  
EMBO Journal  
American Journal of Human Genetics  
Human Molecular Genetics  
American Journal of Medical Genetics  
Trends in Genetics  
European Journal of Human Genetics  
Clinical Genetics  
Journal of Medical Genetics  
Human Genetics

Nucleic Acids Research  
Journal of Cell Biology  
Molecular and Cellular Biology  
Journal of Molecular Biology  
Molecular and Biological Evolution  
Journal of Molecular Evolution  
Somatic Cell and Molecular Genetics  
Cytogenetics and Cell Genetics  
Genomics  
Genome Research  
Mammalian Genome  
Chromosoma  
Chromosome Research  
Gene  
Genes and Development  
Development  
Developmental Dynamics  
Molecular Therapy

### **Professional awards and special recognitions:**

1982 – 1984	Basil O'Connor Award, March of Dimes Birth Defects Foundation
1983 – 1988	Scholar, Medical Research Council of Canada
1988 – 1989	Scientist, Medical Research Council of Canada
1998 – 2003	Franklin Delano Roosevelt Investigator, March of Dimes Birth Defects Foundation
1999	Pruzansky Lecturer, American College of Medical Genetics and March of Dimes Birth Defects Foundation
2001	Outstanding Faculty Award, Biomedical Sciences Training Program, Case Western Reserve University School of Medicine
2001	President, American Society of Human Genetics

### **Organizations and participation:**

2002 – present	Advisory Board, Genetics Prize, Peter Gruber Foundation
2001 – 2002	Board of Directors, Cleveland BioEnterprise Corporation (“BioEnterprise”), Cleveland, OH
2001 – present	Professional Advisory Board, International Rett Syndrome Association
2001 – 2002	Board of Trustees, Great Lakes Science Center, Cleveland, OH
2000 – present	Strategic Advisory Board, NineSigma, Inc., Cleveland, OH
1999 – present	Technology Advisory Council, Biomec, Inc., Cleveland, OH
1999 – 2002	Board of Trustees, Edison Biotechnology Center, Inc.

1995 – present	Co-Founder and Member, Scientific Advisory Board, Athersys, Inc., Cleveland, OH
1988 – 1998	Consultant, Oncor, Inc. and OncorMed, Inc., Gaithersburg, MD

## **Invited Seminars and Lectures (1988-present)**

### **1988**

Invited Speaker, Canadian College of Medical Genetics Symposium on X Chromosome, Montreal, Quebec, February.

Seminar, Department of Genetics, Stanford University, Stanford, CA, March.

Seminar, Department of Human Genetics, University of Michigan, Ann Arbor, MI, April.

Seminar, Howard Hughes Medical Institute, University of California, Los Angeles, CA, May.

Invited Speaker, Great Lakes Chromosome Conference, Toronto, Ontario, May.

Invited Speaker, Symposium on Human Genome Organization, 16th International Congress of Genetics, Toronto, Ontario, August.

Invited Speaker, European Workshop on Automated Chromosome Analysis, Llangollen, North Wales, September.

Seminar, Department of Human Genetics, University of Leiden, The Netherlands, September.

Invited Speaker, Symposium on Gene Expression and Evolution, Queen's University, Kingston, Ontario, October.

Seminar, Genetics Program, University of Iowa, Iowa City, December.

### **1989**

Invited Speaker, International Meeting on Aneuploidy, Reno, NV, January.

Invited Speaker, Royal Canadian Institute, Toronto, Ontario, January.

Seminar, Genetics Research Colloquium, Stanford University, Stanford, CA, February.

Seminar, Division of Genetics, University of Rochester Medical Center, Rochester, NY, March.

Invited Speaker, Symposium on Molecular Approaches to the Human Genome, Tokyo, Japan, March.

Seminar, Program in Genetics, Department of Biochemistry, Emory University, Atlanta, GA, April.

Invited Speaker, Tenth International Chromosome Conference, Uppsala, Sweden, June.

Seminar, Department of Medical Genetics, Mayo Clinic, Rochester, MN, August.

Invited Speaker, American Society for Cell Biology Summer Conference, Chromosome Structure and Segregation, Airlie, VA, September.

Speaker and Co-organizer, Banbury Conference on Molecular Cytogenetics, Cold Spring Harbor, NY, October.

Invited Speaker, National Down Syndrome Society Conference, New York, NY, December.

Invited Speaker, X Chromosome Workshop, Baylor College of Medicine, Houston, TX, December.

### **1990**

Seminar, Department of Physiology, University of California, San Francisco, CA, January.

Seminar, Department of Microbiology and Molecular Genetics, New Jersey Medical School, Newark, NJ, March.

Seminar, Department of Human Genetics, Yale University, New Haven, CT, April.

Invited Speaker, Molecular Cytogenetics Symposium, University of Connecticut, Storbridge, MA, April.

Seminar, Institute for Molecular Genetics, Baylor College of Medicine, Houston, TX, October.

Speaker and Co-organizer, Workshop on X Chromosome Inactivation, Annual Meeting of American Society of Human Genetics, Cincinnati, OH, October.



## 1991

Invited Speaker, X chromosome workshop, Oxford University, Oxford, UK, January.  
Invited Speaker, American Cytogenetics Conference, Lake Tahoe, CA, February.  
Seminar, Department of Pediatrics, University of Colorado Health Sciences Center, Denver, CO, March.  
Seminar, Department of Genetics, Case Western Reserve University, Cleveland, OH, June.  
Invited Speaker, FASEB Summer Conference, Molecular Genetics, Vermont, July.  
Invited Speaker, Gordon Conference, Human Molecular Genetics, Newport, RI, July.  
Invited Speaker, Workshop on X Chromosome Inactivation, Eighth International Congress of Human Genetics, Washington, DC, October.  
Seminar, Department of Radiation Oncology, Stanford University, Stanford, CA, October.  
Seminar, Department of Genetics, University of Washington, Seattle, WA, November.  
Seminar, Department of Biochemistry, Emory University School of Medicine, Atlanta, GA, December.

## 1992

Seminar, Department of Human Genetics, University of Pennsylvania, Philadelphia, PA, January.  
Speaker and Co-organizer, International Symposium on Molecular Cytogenetics, Tahoe, NV, February.  
Invited Speaker, Gordon Conference, Molecular Cytogenetics, Ventura, CA, March.  
Invited Speaker, Markey Symposium on the Genome, University of California, Berkeley, CA, March.  
Seminar, Department of Biochemistry, University of Texas Southwestern School of Medicine, Dallas, TX, April.  
Invited Speaker, Gordon Conference, Biological Mechanisms, Holderness, NH, June.  
Invited Speaker, Annual Meeting of American Cytogenetics Technologists, Rochester, MN, June.  
Invited Speaker, Clinical Genetics Conference, March of Dimes Birth Defects Foundation, Palo Alto, CA, July.  
Invited Speaker, Symposium, Annual Meeting of Canadian College of Medical Genetics, Ottawa, Ontario, September.  
Seminar, Department of Pediatrics, Case Western Reserve University School of Medicine, Cleveland, OH, October.  
Seminar, Ireland Cancer Center, University Hospitals of Cleveland, Cleveland, OH, October.

## 1993

Invited Speaker, Fifth International Conference on Chromosomes in Solid Tumors, Tucson, AZ, January.  
Invited Speaker, Horizons in Biomedical Research, Cleveland Clinic Foundation, Cleveland, OH, January.  
Invited Speaker, Conference on Epigenetic Factors in Inheritance, National Institutes of Health, Bethesda, MD, April.  
Invited Speaker and Co-organizer, Third International Workshop on the X Chromosome, St. Louis, MO, May.  
Visiting Faculty, University of Pittsburgh, Medical Genetics 1993 Review Course, Pittsburgh, PA, May.  
Invited Speaker, "DNA and Chromosomes", Cold Spring Harbor Symposium on Quantitative Biology, Cold Spring Harbor, NY, June.  
Invited Speaker, Workshop on Molecular Mechanisms of Chromosome Abnormalities, American Society of Human Genetics annual meeting, New Orleans, LA, October.  
Invited Speaker, Conference on Epigenetic Modification of the Genome, American Society for Biochemistry and Molecular Biology, Keystone, CO, October.

Invited Speaker, Symposium on Molecular Cytogenetics, American Society of Agronomy Annual Meeting, Cincinnati, OH, November.  
Invited Speaker, Riken International Symposium, "Frontiers of the Human Genome and Biomedical Implications", Tokyo, Japan, November.  
Keynote Speaker, New York Genetics Task Force Annual Meeting, New York, NY, December.  
Invited Speaker, Fourth Annual Miami Children's Hospital Research Institute Symposium, Miami, FL, December.

## 1994

Invited Speaker, 16th Annual Lorne Conference, "Organization and Expression of the Genome", Lorne, Australia, February.  
Seminar, The Murdoch Institute, Royal Children's Hospital, Melbourne, Australia, February.  
Invited Speaker, Symposium on "The Human Genome: Where is it Taking Us?", University of Chicago, March.  
Seminar, Frontiers in Cell Biology, Columbia University College of Physicians and Surgeons, New York, NY, May.  
Zeller Visiting Lecturer in Family Medicine, St. Elizabeth Hospital Medical Center, Youngstown, OH, September.  
Seminar, Department of Genetics, Washington University School of Medicine, St. Louis, MO, November.  
Seminar, Lineberger Cancer Center, University of North Carolina, Chapel Hill, NC, December.  
Invited Speaker, Association of Chairmen of Departments of Physiology Annual Meeting, San Jose Del Cabo, Mexico, December.

## 1995

Seminar, Department of Genetics, University of Pennsylvania School of Medicine, Philadelphia, PA, January.  
Keynote Speaker, GSBS Program in Genetics Symposium, University of Texas-Health Science Center, Houston, TX, January.  
Seminar, Department of Chemistry, Cleveland State University, Cleveland, OH, February.  
Seminar, National Center for Human Genome Research, National Institutes of Health, Bethesda, MD, February.  
Invited Speaker, Keystone Symposium on Molecular Basis for Differences Between the Sexes, Tamarron, CO, February.  
Visiting Professor, University of Maryland at Baltimore, Division of Human Genetics, Baltimore, MD, April.  
Invited Speaker, Symposium on "The Year in Genetics," The Clinical Research Meeting, San Diego, CA, May.  
Visiting Faculty, Educational Conference on "Applying Molecular Genetics to Cancer Detection and Management," Washington, DC, September.  
Invited Speaker, NIEHS Workshop on Aneuploidy: Etiology and Risk Factors, Research Triangle Park, NC, September.  
Seminar, Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX, October.  
Invited Speaker, American Association for the Study of Liver Diseases, Postgraduate course on "Genetic and Molecular Advances in Liver Disease," Chicago, IL, November.  
Seminar, Genetics Laboratories, Department of Biochemistry, University of Oxford, Oxford, UK, December.

## 1996

Seminar, Institute of Human Genetics, University of Minnesota, Minneapolis, MN, January.  
Invited Speaker, Symposium on Genomic Imprinting, Third Joint Clinical Genetics Meeting, San Antonio, TX, March.  
Invited Speaker, Genetics Symposium, Duke University, Research Triangle, NC, April.  
Invited Speaker, Cambridge Symposium on Nuclear Structure, Bolton Valley, VT, May.  
Seminar, Sloan-Kettering Cancer Center, New York, NY, June.  
Seminar, Department of Human Genetics, Mt. Sinai School of Medicine, New York, NY, June.  
Invited Plenary Speaker, 9th International Congress of Human Genetics, Rio de Janeiro, Brazil, August.  
Seminar, Human Genetics Program, University of Massachusetts, Worcester, MA, September.  
Invited Speaker, Juan March Foundation Workshop on Chromosome Behaviour, Madrid, September.  
Invited Speaker, Symposium on Genetics in Development, Evolution and Disease, Wayne State University School of Medicine, Detroit, MI, October.  
Co-Organizer and Speaker, Workshop on Centromeres and Telomeres, Human Genome Organization, Oxford, UK, October  
Invited Speaker, Workshop on Chromosome Abnormalities, American Society of Human Genetics Annual Meeting, San Francisco, October.  
Seminars, Western General Hospital and University of Edinburgh, Edinburgh, Scotland, December.  
Invited Speaker, Workshop on Artificial Human Chromosomes, Milan, Italy, December.

## 1997

University Lecturer, University of Texas Southwestern Medical Center, Dallas, TX, January.  
Seminar, Biomedical Sciences Program, University of California at San Francisco, San Francisco, CA, January.  
Seminar, Interdepartmental Genetics Program, Iowa State University, Ames, IA, April.  
Keynote Speaker, Great Lakes Area Regional Genetics Group, Annual Meeting, Cleveland, OH, April.  
Seminar, Genzyme Genetics, Inc., Framingham, MA, May.  
Seminar, Department of Genetics, Yale University, New Haven, CT, June.  
Invited Speaker, Gordon Conference on Epigenetics, Holderness, NH, August.  
Speaker and Moderator, Gene Therapy Policy Conference, National Institutes of Health, Bethesda, MD, September.  
Invited Speaker, Conference on Human Gene Therapy: The Next Generation, Paris, September.  
Seminar, Program in Genetics, Northwestern University, Chicago, IL, October.  
Invited Speaker, Symposium on "Beyond the Sequence of the Human Genome", Council of Academic Societies Plenary Session, American Association of Medical Colleges Annual Meeting, Washington, DC, November.  
Seminar, Howard Hughes Medical Institute, University of Iowa, Iowa City, IA, November.  
Seminar, Program in Genetics, University of Iowa, Iowa City, IA, November.  
Seminar, Department of Genetics, Yale University, New Haven, CT, November.

## 1998

Seminar, Program in Cellular and Molecular Medicine, University of California, San Diego, San Diego, CA, January.  
Seminar, Salk Institute, San Diego, CA, January.  
Invited Speaker, Institute for Genetic Medicine Symposium, "Genomic Genetics", University of Southern California, Pasadena, CA, January.

Invited Speaker, Miami Winter Symposium, “Advances in Gene Technology”, Miami, FL, February.

Seminar, Department of Molecular Genetics, University of Cincinnati College of Medicine, Cincinnati, OH, February.

Invited Faculty, Course on Genetics in Medicine, American Medical Association, New Orleans, LA, March.

Seminar, Department of Zoology, Miami University, Oxford, OH, April.

Seminar, Department of Molecular Biotechnology, University of Washington, Seattle, WA, May.

Invited Speaker, Workshop, American Society of Gene Therapy, Seattle, WA, May.

Invited Speaker, Summer Symposium, American Association of Medical Colleges, San Francisco, CA, July.

Invited Speaker, Molecular Genetics Gordon Conference, Newport, RI, July.

Visiting Faculty, 39th Annual Short Course in Medical and Experimental Mammalian Genetics, Bar Harbor, ME, July.

Invited Speaker, Curie Institute Symposium on Epigenetics and DNA Methylation, Paris, France, September.

Invited Speaker, Fourth Annual Pauline Wilson Horner Genetics Symposium, Cleveland, OH, October.

Invited Speaker, International Workshop on the Human Genome, Valencia, Spain, October.

Invited Speaker, Workshop on Artificial Chromosomes, American Society of Human Genetics Annual Meeting, Denver, CO, October.

Seminar, Genetics Program, MD Anderson Cancer Center, Houston, TX, November.

Seminar, Institute for Human Gene Therapy, University of Pennsylvania, December, Philadelphia.

## 1999

Invited Speaker, Frontiers at the Millennium, International Society for Advancement of Humanistic Studies in Medicine, Crested Butte, CO, February.

Presidential Visiting Scholar, Chico State University, Chico, CA, March.

Medical Grand Rounds, Enloe Medical Center, Chico, CA, March.

Pruzansky Lecturer, Annual Meeting of the March of Dimes and American College of Medical Genetics, Miami, FL, March.

Invited Speaker, Group on Resident Affairs annual meeting, Association of American Medical Colleges, Monterey, CA, April.

Seminar, Department of Human Genetics, University of Chicago, Chicago, IL, May.

Invited Speaker, Council of Teaching Hospitals annual meeting, Association of American Medical Colleges, West Palm Beach, FL, May.

Seminar, Genomics Group, Fred Hutchinson Cancer Center, Seattle, WA, June.

Invited Speaker, Symposium on Chromatin Structure and Gene Regulation, Annual Meeting of The Endocrine Society, San Diego, CA, June.

Seminar, Pasteur Institute, Paris, France, June.

Invited Speaker, European Cytogenetics Conference, Vienna, Austria, July.

Visiting Faculty, 40th Annual Short Course in Medical and Experimental Mammalian Genetics, Bar Harbor, ME, July.

Invited Speaker, Human Molecular Genetics Gordon Conference, Newport, RI, August.

Invited Speaker, Genetics Symposium, Emory University School of Medicine, Atlanta, GA, September.

Invited Speaker, Frontiers in Clinical Genetics, George Washington University Medical Center, Washington, DC, November.

Seminar, Human Medical Genetics Program, University of Colorado Health Sciences Center, Denver, CO, December.

## 2000

Seminar, Department of Human Genetics, University of California, Los Angeles, CA, February.  
Invited Speaker, Tenovus Symposium, Glasgow, Scotland, April.  
Invited Speaker, Rett Syndrome Conference, Washington, DC, June.  
Keynote Speaker, Genetics in the 21<sup>st</sup> Century Conference, Columbus, OH, June.  
Invited Speaker, Gene Therapy Research Symposium, March of Dimes Conference, Matsue, Japan, July.  
Visiting Faculty, 41st Annual Short Course in Medical and Experimental Mammalian Genetics, Bar Harbor, ME, July.  
Invited Speaker, Department of Pediatrics, University of Illinois at Chicago Medical Center, Chicago, IL, July.  
Invited Speaker, NIH Conference on Klinefelter Syndrome, Washington, DC, August.  
Seminar, Department of Medical and Molecular Genetics, Indiana University, Indianapolis, IN, September.  
Invited Speaker, Molecular Genetics Update for the Endocrinologist, Genes 2000 Pediatric Endocrinology International Congress, Sydney, Australia, October.  
Seminar, Division of Human Genetics, Washington University School of Medicine, St. Louis, MO, November.  
Seminar, Department of Genetics, Duke University School of Medicine, Durham, NC, December.

## 2001

Seminar, Division of Medical Genetics and Markey Molecular Medicine Program, University of Washington, Seattle, WA, January.  
Invited Speaker, AAAS Genome Symposium, San Francisco, CA, February.  
Plenary Speaker, 2<sup>nd</sup> Annual Conference on Sex and Gene Expression, Winston-Salem, NC, March.  
Seminar, Edinburgh Chromatin Group, Edinburgh, Scotland, April.  
Invited Speaker, Symposium on Gene Regulation, HGM2001 Conference, Edinburgh Scotland, April.  
Invited Speaker, Gordon Conference, Human Molecular Genetics, Newport, RI, August.  
Seminar, Division of Human Genetics, University of Cincinnati, Cincinnati, OH, September.  
Invited Speaker, Nordic Association for Andrology Annual Meeting, Symposium on Klinefelter Syndrome, Copenhagen, Denmark, October.

## 2002

Invited Speaker, Society for Women's Health Annual Conference, Stanford, CA, February.  
Invited Speaker, International Society of Barristers, Kona, Hawaii, March.  
Keynote Speaker, Annual Scientific Retreat, Children's Hospital Research Institute, Columbus, OH, April.  
Invited Speaker, 3<sup>rd</sup> Annual Rett Syndrome Symposium, Rett Syndrome Research Foundation, Baltimore, MD, June.  
Panelist, Summit for a Cure, MSNBC special broadcast (hosted by Brian Williams), Cleveland, OH, June.

## Teaching responsibilities including continuing education:

### Graduate

1982

MBP 1012-1014L - 'Molecular Organization of the Human Genome', Department of Medical Genetics, University of Toronto (12 hrs)

1987	MBP 1014L - 'Human Molecular Genetics', Department of Medical Genetics, University of Toronto (6 hrs of 24 hr course, with Drs. R. Worton, L. Tsui and D. Cox)
1991	GEN 210 - 'Advanced Human Genetics', Department of Genetics, Stanford University (with Dr. D. Botstein)
1992 – 2002	GENE 500/504 - 'Advanced Eukaryotic Genetics', Department of Genetics, Case Western Reserve University (with other faculty)
1995, 1997	GENE 515 - 'Chromosome Structure and Function', Department of Genetics, Case Western Reserve University
1996, 1998, 2000	GENE 510 - 'Advanced Human Genetics', Department of Genetics, Case Western Reserve University
1997, 1998	C3MB - 'Cell and Molecular Biology', Human Genetics Section, Case Western Reserve University (with Dr. M. Warman)
2002	GENE 512 – 'Analysis of Complex Genomes', Department of Genetics, Case Western Reserve University

#### Medical Students

1983 - 1988	Course Director, Genetics for Second Year Medical Students (30 hrs), Faculty of Medicine, University of Toronto
1982 - 1988	Lecturing in Genetics for Second Year Medical Students, University of Toronto (10-18 hrs)
1989 - 1992	Lecturing in Genetics 201, Stanford University (4 - 10 hrs)
1990 - 1992	Course Director, Genetics 201 (36 hrs), Stanford University
1992 - 1996	Lecturing in First Year Committee course for medical students, Case Western Reserve University School of Medicine (~5 hrs)

#### Undergraduate

1985 - 1987	MGB 470H - "Cellular and Molecular Mammalian Genetics", Faculty of Arts and Sciences, University of Toronto (6-10 hrs)
1987 - 1989	MGB 470H - "Molecular Genetics of Complex Genomes", Faculty of Arts and Sciences, University of Toronto (26 hr course with Dr. L. Tsui)
1993 - 1996	BIO 214 - "Genetics", School of Arts and Sciences, Case Western Reserve University (2 hrs)

### **Graduate Students Supervised**

#### University of Toronto

1982 - 1985	Leslie A. Creeper, M.Sc. Thesis: <i>Functional analysis of a mammalian chromosomal origin in yeast</i>
1984 - 1987	John S. Wayne, Ph.D. Thesis: <i>Organization and evolution of chromosome-specific human alpha satellite DNA</i>
1984 - 1990	Sharon J. Durfy, Ph.D. Thesis: <i>Nucleotide sequence variation, homogenization, and evolution of X chromosome alpha satellite DNA</i>
1987 - 1990	Carolyn J. Brown, Ph.D. Thesis: <i>Studies of human X chromosome inactivation</i>
1987 - 1992	Rachel Wevrick, Ph.D. Thesis: <i>Organization and behavior of alpha satellite DNA at the centromeres of human chromosomes</i>
1986 - 1993	Peter E. Warburton, Ph.D.

Thesis: *Evolution of tandemly repeated DNA: repeat unit variation of human alpha satellite DNA*

Stanford University

- 1988 - 1993      Melanie M. Mahtani, Ph. D.  
                          Thesis: *Physical and genetic mapping studies of the human X chromosome: repression of recombination at the centromere*
- 1990 - 1995      Laura Carrel, Ph.D.  
                          Thesis: *Developmental and chromosomal basis of gene regulation on the mammalian X chromosome*
- 1991 - 1995      Brian Hendrich, Ph.D.  
                          Thesis: *Characterization of the human XIST gene and its promoter: implications for mammalian X chromosome inactivation*
- 1991 - 1995      Andrew Miller, Ph.D.  
                          Thesis: *Physical mapping studies and X inactivation analysis on the short arm of the human X chromosome*

Case Western Reserve University

- 1994 – 1998      Robert Plenge, M.D., Ph.D.  
                          Thesis: *Genetic control of skewed X chromosome inactivation*
- 1995 – 2000      Anne W. Higgins, Ph.D.  
                          Thesis: *Centromere structure and behavior: insight from engineered dicentric chromosomes*
- 1996 – 2001      Mary G. Schueler, Ph.D.  
                          Thesis: *Structure and evolution of the pericentromeric region of the human X chromosome*
- 1998 – 2002      Ivona Percec, M.D., Ph.D. [jointly with University of Pennsylvania; Marisa Bartolomei, advisor]  
                          Thesis: *Genetic dissection reveals multiple autosomal elements involved in X chromosome inactivation in the mouse*
- 1997 – present      James Amos-Landgraf (to obtain Ph.D.)
- 1999 – present      M. Katharine Rudd (to obtain Ph.D.)
- 1999 – present      Lisa Helbling Chadwick (to obtain Ph.D.)
- 2000 – present      Cory Valley (to obtain Ph.D.)

**Postdoctoral Fellows / Research Associates / Senior Research Associates Supervised**

- 1988 - 1991      Cecil B. Sharp, D.M.D., Ph.D.
- 1990 - 1992      Thomas Haaf, M.D., Ph.D.
- 1991 - 1994      Tiina Alitalo, Ph.D.
- 1991 - 1994      Carolyn J. Brown, Ph.D.
- 1991 - 1992      Cordula Kirchgessner, Ph.D.
- 1992 - 1994      Dayna Wolff, Ph.D.
- 1994 - 1997      John Harrington, Ph.D.
- 1994 - 1996      Gil van Bokkelen, Ph.D.
- 1994 - 1998      Kosuke Sakai, Ph.D.
- 1995 - 1996      Beth Sullivan, Ph.D.
- 1995 - 1999      Laura Carrel, Ph.D.
- 1996 - 1998      R. Willie Mays, Ph.D.
- 1996 – 2000      Karen Tsuchiya, M.D.

1998 – 2001	Christie Gunter, Ph.D.
1999 – 2003	Brian P. Chadwick, Ph.D.
1999 – present	Brenda Grimes, Ph.D.
2000 – present	Kristin C. Scott, Ph.D.
2000 – present	Satkunanathan (Bala) Balakumaran, Ph.D.

**Areas of research interests (basic and applied) – list:**

Human Genetics	Genome Organization
Chromosome Structure and Function	X-Linked Disease
Epigenetics	Gene Therapy